# Joubert Syndrome

## Main Features

- Cerebellar vermis hypoplasia or aplasia
- episodic tachypnea (“panting like a dog”) and apnea in infancy
- jerky eye movements, hypotonia, developmental delay

## Eye Findings

- **Ocular Motor Abnormalities**
  - Absent smooth pursuit
  - Hypometric saccades with prolonged latency; they change direction of fixation by turning their heads
  - Nystagmus – pendular, rotary, horizontal, see-saw, torsional
- **Congenital Retinal Dystrophy** (in majority of patients; some definitely have normal appearing fundi)
  - Progressive chorioretinal pigmentary changes including reports of “mottling” in periphery
  - Chorioretinal Coloboma
  - Attenuation of retinal arterioles
  - Nonrecordable / attenuated ERG, but Preserved flash and pattern VEPs (Differentiates from LCA)
  - Rod photoreceptors are more severely affected than cones
  - May have good Best Corrected Visual Acuity – reports of 20/40 (also better than LCA)
  - Those with Retinal dystrophy also have Renal cysts and other kidney disease
- Ptosis – Bilateral or Unilateral
- Strabismus
- Supranuclear Ocular motor palsies

### Differentiating Joubert’s from Leber’s Congenital Amaurosis

- Both have nonrecordable or severely attenuated ERG, but Joubert’s has recordable (less than normal) flash and pattern VEPs
- Joubert’s generally has better Visual acuity

## Etiology

- Autosomal Recessive
- First described in 1969, by 1991, 94 patients reported
- More commonly reported in cultures of consanguinity, although distributed worldwide

## Other Findings

- Neuro: Mental retardation, ataxia
- Respiratory: Episodic tachypnea and apnea that improves with age
- Renal: Renal cysts, inflammation, sclerosis, can be deadly
- GI: protruding tongue, tongue tumors, pyloric stenosis, duodenal atresia, hepatic inflammation
- Bone: Polydactyly

## References


- Lambert, et al. Arch Ophth 1989;107:709-713
- Sztriha, et al. Ped Neurol 1999; 20:274-281
- Saraiva, Am J of Med Genetics 1992; 43:726-731
- OMIM 213300, 608091


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